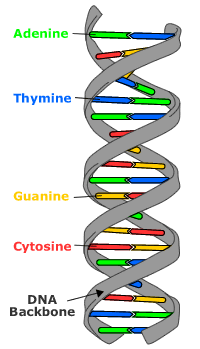
Name: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Date: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ Period: \_\_\_\_\_

**Unit 6, Part 4 Notes – Mutations**

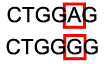
Ms. Ottolini, Pre-AP Biology

**What is a mutation and who cares?**

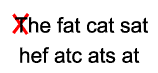
1. A mutation is a change in the sequence of \_\_\_\_\_\_\_\_\_\_\_ \_\_\_\_\_\_\_\_\_\_\_\_(A, T, C, and G’s) in the DNA code.
2. Recall from our study of protein synthesis that the sequence of bases in DNA ultimately codes for sequences of \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ found in protein. Therefore, any change in the DNA sequence will affect the resulting polypeptide.
3. Examples:

* ***Sickle Cell Disease:*** A mutation in the gene coding for the hemoglobin protein causes an abnormally shaped protein that causes red blood cells to form a crescent moon shape. These abnormally shaped red blood cells can clog arteries.
* ***Tay-Sacchs Disease****:* A mutation in the gene coding for an enzyme normally found in lysosomes causes the enzyme to change shape and become dysfunctional. Normally, the enzyme’s substrate is a fatty substance called GM2 ganglioside, which it helps to break down. If the enzyme’s active site changes shape, it can no longer bind GM2. When left unbroken, GM2 can build up in the brain and spinal tissue. This build-up impairs nerve function and causes death by approximately age 4

**Types of Mutations – Base Sequence Changes**

1. There are two types of mutations based on their effect on the resulting polypeptide: point mutations and frameshift mutations.
2. ***Point Mutations:*** these mutations are caused by a \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ in the DNA sequence (one base is exchanged for another)

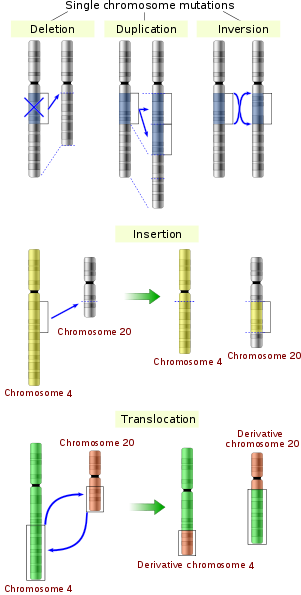
* ***\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_****:* If the DNA change produces an mRNA codon that codes for the same amino acid as the original sequence, there will be no effect on the resulting polypeptide
* ***\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_****:* If the DNA change produces an mRNA codon that codes for a different amino acid as the original sequence, there will be a change in one amino acid in the resulting polypeptide.
* ***\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_****:* If the DNA change produces an mRNA codon that does not match with any amino acid (i.e., the stop codons UAA, UAG, and UGA), then the creation of the polypeptide will stop.

1. ***Frameshift Mutations:*** these mutations are caused by an \_\_\_\_\_\_\_ or \_\_\_\_\_\_ of bases in the DNA sequence. Since DNA codes for mRNA that is divided into codons that are three bases long, insertions and deletions in the DNA sequence can alter a gene so that its message is no longer correctly “grouped.” If there is a frameshift mutation early on in a protein-coding DNA sequence, all amino acids created after the mutation will change.

For example, consider the sentence, “The fat cat sat.” Each word represents a codon. If we delete the first letter but still arrange the letters in groups of three, the sentence no longer “makes sense.” It reads, “Hef atc ats at.”

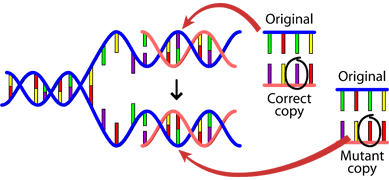
**Types of Mutations – Large Chromosomal Mutations**

1. Mutations that involve changes in large “chunks” of a chromosome are called chromosomal mutations. Several types are listed below:
2. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_: A large section of the chromosome is copied.
3. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_: A large section of the chromosome is flipped around.
4. **\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**: A large section from one chromosome is “stuck” into another.
5. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_: Large sections from two chromosomes are switched.



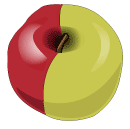
**Causes of Mutations**

1. ***DNA fails to copy accurately:*** Occasionally \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ will make an error when pairing new nucleotides with nucleotides on the template strand of DNA. It may match a C with an A, rather than a T with an A.



1. ***External influences:*** Mutations can also be caused by exposure to specific chemicals or radiation that are called \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_. These factors cause the DNA to break down. This is not necessarily unnatural – even in the most isolated, perfect environment, DNA breaks down. The cell has enzymes that are used to repair DNA. However, if these enzymes do not perfectly repair the errors caused by mutagens, a true mutation results.

**Can Mutations be Passed Down to Offspring?**

1. Since all cells in our body contain DNA, there are a lot of places for mutations to occur ; however, some mutations cannot be passed on to offspring.
2. ***\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_*** occur in normal body cells (non-reproductive cells) and won’t be passed on to offspring. For example, the golden color on half of this Red Delicious apple was caused by a somatic mutation. Its seeds will not carry the mutation.
3. ***\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_*** occur in gametes (eggs and sperm). These mutations can be passed on to offspring.

|  |  |  |
| --- | --- | --- |
|  | |  | | --- | |  | |

**Are all mutations “bad”?**

1. Not all mutations are bad, and some may have more effects than others on an individual’s traits. Besides negative mutations that can cause diseases (discussed above), there are also neutral and positive mutations.
2. ***\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_***: If a mutation codes for the same amino acid (silent mutation) it will not have an effect on an organism’s traits. Also, if a mutation located in a section of DNA that is not used to code for a protein, it will similarly have no effect on an organism’s traits.
3. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_: A mutation may allow an organism to produce a protein that is beneficial to that organism. For example, certain bacteria have a mutation that allows them to produce a protein to break down antibiotics. This makes them resistant to the effects of antibiotics.